

LSM11 gene

LSM11, U7 small nuclear RNA associated

Normal Function

The *LSM11* gene provides instructions for making one piece (subunit) of a group of proteins called the U7 small nuclear ribonucleoprotein (snRNP) complex. In cells, the U7 snRNP complex plays an important role in processing molecules called messenger RNA (mRNA), which serve as the genetic blueprints for making proteins. The mRNA starts out as a molecule called pre-mRNA and must go through several processing steps before it is ready to make a protein. The U7 snRNP complex helps to process pre-mRNA. Specifically, the U7 snRNP complex targets the pre-mRNA of proteins called histones. Histones are structural proteins that attach (bind) to DNA and are necessary for copying a cell's DNA before the cell divides (a process known as DNA replication).

Health Conditions Related to Genetic Changes

Aicardi-Goutières syndrome

Variants (also called mutations) in the *LSM11* gene have been found to be a rare cause of Aicardi-Goutières syndrome, a disorder that typically involves severe brain dysfunction (encephalopathy), skin lesions, immune system abnormalities, and other health problems.

It is likely that *LSM11* gene variants lead to the production of an abnormal U7 snRNP complex that cannot properly process the pre-mRNA of histone proteins. Abnormal processing of histone pre-mRNA can interfere with DNA replication and cause the cell to produce RNA and DNA fragments. Research suggests that these fragments can accumulate and trigger an immune response, which causes the immune system to attack the body's own tissues and organs. This immune response leads to the signs and symptoms of Aicardi-Goutières syndrome.

Other Names for This Gene

- AGS8
- FLJ38273
- U7 SnRNA-Associated Sm-Like Protein LSm11

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LSM11 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=134353\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=134353[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=LSM11+AND+Aicardi-Gouti%C3%A8res+Syndrome>)

Catalog of Genes and Diseases from OMIM

- LSM11, U7 SMALL NUCLEAR RNA-ASSOCIATED PROTEIN; LSM11 (<https://omim.org/entry/617910>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/134353>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LSM11\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LSM11[gene]))

References

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